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Answer to Case of the Month #160 Dysplasia Epiphysealis Hemimelica (Trevor's Disease)

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Clinical Presentation

A 10-year-old boy presented with a 3- to 4-year history of right-sided ankle pain that was aggravated by physical activity and progressed in severity during the day. There was no history of trauma and no other significant medical abnormalities. On examination of the right ankle and foot, the patient had inward pronation and loss of foot arches. Dorsiflexion induced pain, and there was limited motion in the plantar flexion range. Anteroposterior (Figure 1) and oblique (mortise view) (Figure 2) radiographs were obtained.

Imaging Findings

The anteroposterior and medial oblique radiographs (Figures 1, 2) demonstrate a mass-like configuration to the distal fibula that results in mild valgus ankle deformity. Reconstructed coronal and sagittal computed tomographic images (Figures 3, 4) show marked overgrowth of the distal fibula, largely confined to the anteromedial epiphysis, with associated mass effect medially on the minimally deformed tibial plafond and talus.

Diagnosis

Dysplasia epiphysealis hemimelica (Trevor's disease).

Discussion

Dysplasia epiphysealis hemimelica (DEH) is a rare developmental skeletal disorder characterized by osteocartilaginous overgrowth of one or more epiphyses typically in the lower limb [1]. It was first described in 1926 by Mouchet and Berlot [2] who named the condition tarsomegalie. In 1950, Trevor reported 10 cases and referred to this overgrowth as tarsoepiphyseal aclasis [3]. Subsequently, in 1956, Fairbank [1] described an additional 14 cases and coined the term ''dysplasia epiphysealis hemimelica.'' This term reflects the characteristic hemimelic involvement in this condition; typically, either the medial or the lateral side of the epiphysis is affected, with the medial side affected twice as frequently [4–6]. This condition has also been referred to as Trevor's disease.

Involvement of the lower extremity has been divided into 3 different forms. In the localized form, there is involvement of a single bone and typically affects the bones of the hindfoot or ankle [7]. In the classic form, the characteristic hemimelic distribution affects more than one bone in the lower extremity. The talus, distal femoral epiphysis, and distal tibial epiphysis are commonly affected with this variant [7]. The classic form accounts for more than two-thirds of reported cases [7]. Lastly, the generalized or severe form involves the entire lower extremity from the pelvis to the foot or ankle [7]. Involvement of the upper extremity has been reported but is very uncommon [8–10]. Of the 57 cases previously reported in the literature, the 4 most common locations of involvement were the talus-calcaneus (22%), distal tibia-fibula (22%), distal femur (21%), and proximal tibia (11%) [8].

Key Words: Epiphyses; Osteochondroma.

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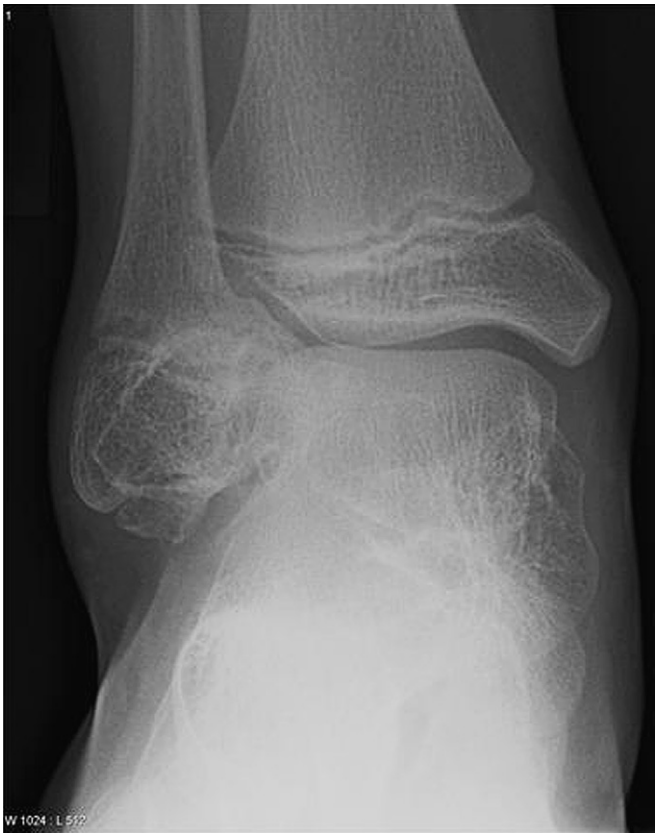

Figure 1. Anteroposterior radiograph of the right ankle.

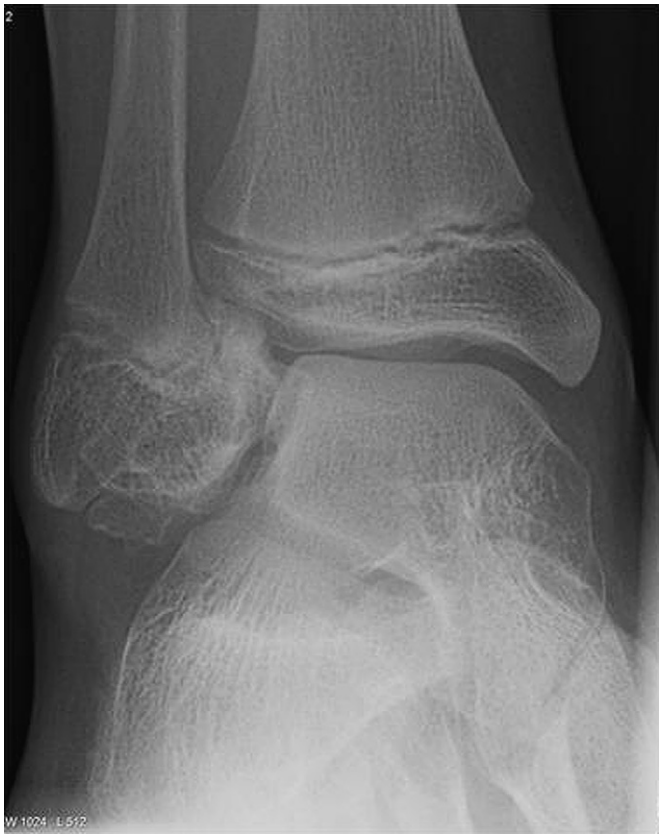

Figure 2. Oblique (mortise view) radiograph of the right ankle.

The incidence of DEH has been commonly cited as 1:1,000,000 [11]. However, it may be higher, because the diagnosis is often unrecognized [4]. Although DEH was described in adults [12], it is primarily a pediatric disease, with the age of onset usually between 2 and 14 years of age [4,6,8]. The disorder is more common in boys than girls, with a reported sex predominance of 3–8:1 [4,13]. Also, the condition usually occurs in whites of northern European descent, with only a few reported cases among blacks [14,15].

The etiology of this condition is unclear [4,8]. It does not appear that hereditary factors play a role in its pathogenesis [13]. It was suggested that an abnormality in the regulation of cartilage proliferation in the affected epiphyses and tarsal bones accounts for the overgrowth [6]. The problem may also be congenital; Fairbank [1] postulated that focal hemorrhage in the limb bud during fetal development results in DEH. On pathologic examination, there is an overgrowth of cartilaginous cells, with scattered enchondral ossification and a cartilaginous cap [4,6,7].

The most common presentation is a painless lump that increases in size [8]. There may also be asymmetric and firm swelling that affects the medial or lateral aspect of the knee or ankle, which will feel bony in consistency, without involvement of the soft tissues [6,8]. Other common complaints include stiffness, leg-length discrepancy, aching pains, and limited range of motion [4,6,15]. Deformity of the joint may result in genu valgum or varum at the knee or equinus deformity of the ankle [15].

There are characteristic plain radiograph abnormalities associated with DEH. Initially, there is an irregular, multicentric, lobulated mass located adjacent to the affected epiphysis or tarsal bone, usually on the medial side [5,15]. There usually is diffuse calcific stippling throughout this mass [7]. The affected ossification center often appears prematurely, with resulting premature ossification of the epiphysis [4,5,16]. As the child ages and the lesion matures, the multicentric mass coalesces and becomes confluent with the underlying bone, forming an irregular, lobulated mass [4–6,15]. During this progression, calcification will become more extensive [7]. Articular surface irregularities may also be present, predisposing to secondary osteoarthritis [6].

Other abnormalities that may be seen by plain radiographs include limb-length discrepancy and deformity that result from premature closure of the physis [4]. Abnormalities separate from the epiphysis, such as metaphyseal spur or exostosis, have also been described [7]. The radiographic differential diagnosis includes myositis ossificans, infection, tumoral calcinosis, synovial osteochondromatosis, and vascular or parasitic calcification [5].

Computed tomography may aid in diagnosis by demonstrating the relationship between the osteocartilaginous

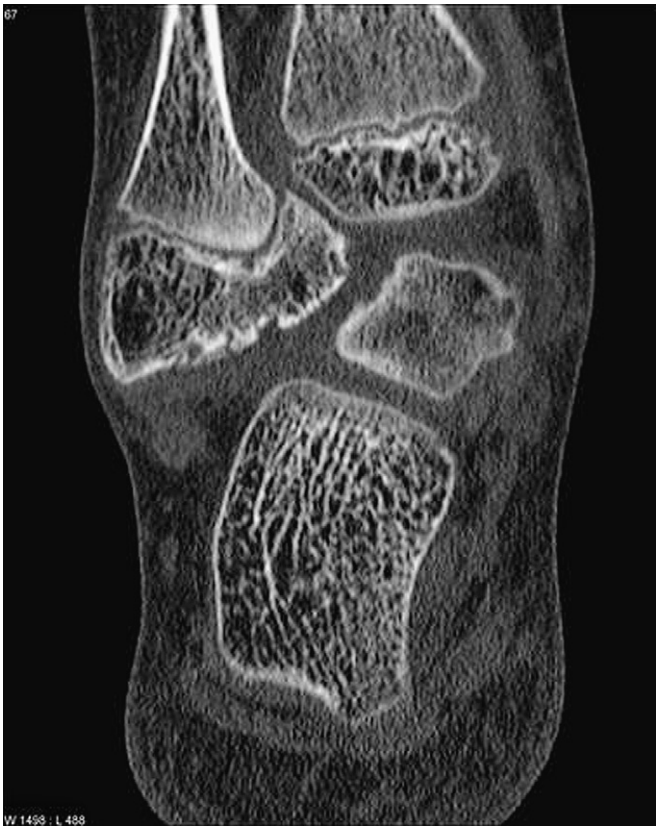

Figure 3. Coronal reconstructed computed tomographic image of the right ankle.

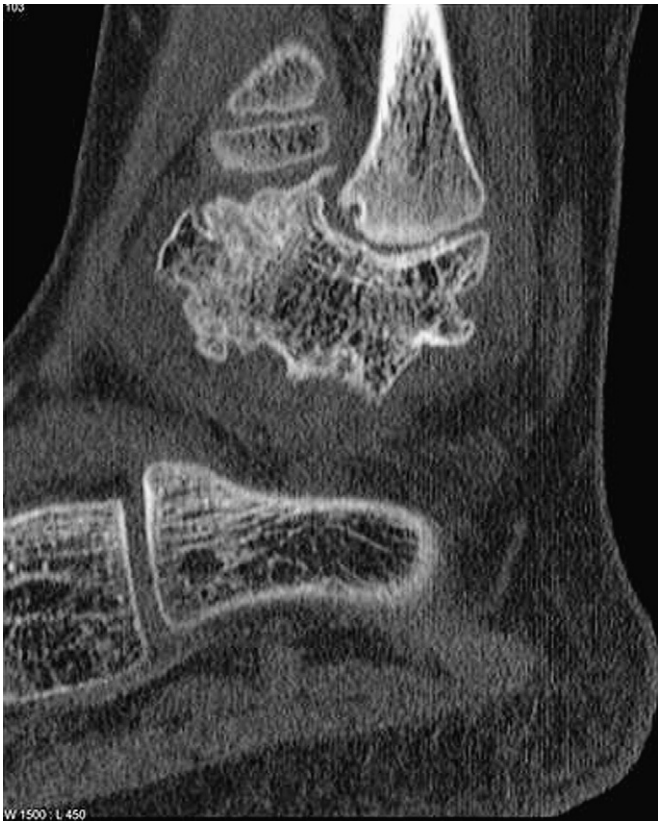

Figure 4. Sagittal reconstructed computed tomographic image of the right ankle.

overgrowth, bone, and soft tissues, in addition to providing information about the integrity of the articular cartilage [8,15,17]. However, assessment of soft tissues and cartilage is limited, and magnetic resonance imaging (MRI) is the next most useful imaging modality after plain radiography. The effects of the epiphyseal overgrowth and resulting altered joint mechanics on surrounding ligaments, tendons, and cartilage, including menisci, can be accurately assessed by using MRI [18]. T2* gradient-echo imaging accurately reveals bony anatomy, which can be further enhanced by 3-dimensional volume acquisitions and fat-suppressed sequences, the latter to assess for bone marrow oedema. Patients with ankle involvement often have bony oedema because of the high mechanical stress of ambulation on the foot and ankle from altered joint alignment and/or mechanics [18]. This oedema is usually patchy and present in the overgrown and normal bone components, and must be distinguished from the focal linear abnormalities characteristic of stress fracture [18].

The ability of MRI to detect chronic tendonitis, tenosynovitis, and ligamentous abnormalities that result from abnormal bony pressure and/or joint mechanics may direct therapeutic measures to the specific bony prominence responsible for symptoms [18]. MRI may also aid in diagnosis if there are only a few small or discrete calciferous foci instead of the classic radiographic presentation [19]. In these cases, MR can be used to exclude calcified and/or ossified para-articular tumour [19].

Once the initial diagnosis of DEH is made, the presence of other sites of involvement should be considered, because more than one site is usually affected [7]. Surveillance should be undertaken until puberty, because the lesions may not all present simultaneously [7]. These other areas of involvement may be detected through appropriate clinical history and physical examination, with targeted plain radiographs obtained when necessary.

Treatment should be undertaken if the patient is symptomatic with pain, joint deformity, or limited range of motion [15]. If the lesion is extra-articular, then surgical excision is recommended, and excellent results are usually attained [4,8,15]. Excision of intra-articular lesions is associated with poorer outcomes and may lead to premature osteoarthritis [8]. It is generally discouraged, unless the lesion becomes a loose body [4]. In cases in which an intra-articular mass causes angular deformity and the articular surface is smooth, extra-articular osteotomy may be undertaken to correct the deformity [15]. Recurrence can be predicted after osteotomy if the growth plate at the affected joint is open and active [15]. In asymptomatic cases, the patients may be observed, because no cases of malignant transformation have been reported [4].

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